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Positive and Negative Experiences of Parenting a Pre-school Child with 22q11.2 Deletion Syndrome.

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Cover Page

"You don't know until you get there":

The positive and negative 'lived' experience of parenting an adult child with 22q11.2 deletion syndrome.

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Abstract

Objectives: 22q11.2 deletion syndrome (22q11DS), a complex phenotype associated with more than 180 features, presents complex challenges for parents including gaining an accurate diagnosis. This phenomenological study sought the 'lived' interpretations of parents supporting an adult child with 22q11DS, a poorly researched area.

Method: Interpretative Phenomenological Analysis informed a detailed and open exploration of parenting a child through to adult life with 22q11DS. Using in-depth semi-structured interviews, eight parents (two male, six female) of adult children with 22q11DS were individually interviewed; providing the data set for transcription and thematic analysis.

Results: *Losing 'I' Finding 'self'*, overarched six subordinate themes that emerged from participants' articulated descriptions of psychological distress and psychological growth. Distress in parenting a child with 22q11DS was experienced through stigma, loss, grief, and guilt. Progressively, stigma undermined independence, friendships, and instinctual judgement. Ill-informed hierarchical structures experienced as layers of obstruction and ignorance triggered angry advocacy for their child. Diagnosis brought opposing relief and grief. In time they came to value their unique 'accomplishments', collected on their journey with 22q11DS, and in turn, consciously valued authentic 'self' expressed through empathy, humility, gratitude, and pride.

Conclusion: Unrelenting distress through societal, educational, and health care invalidation persisted for decades for all participants. Conversely, distress facilitated psychological growth for redefining 'self' and role as parents over time. Building on this phenomenological cameo, future research can educate against the plight of 22q11DS families. It can enlighten health care professionals in buffering against associated stigma, blame, and self-doubt, and in fostering psychological wellbeing.

Key words: *Velo-cardio-facial syndrome; 22q11.2 Deletion Syndrome; IPA; traumatic distress; psychological growth*

Introduction

Little is known of the experience of parenting a child with the developmental disability 22q11.2 deletion syndrome (22q11DS; also known as velo-cardio-facial syndrome) despite a prevalence of 1 in 4000 live births (Oskarsdottir, Vujic, & Fasth, 2004). 22q11DS has a complex phenotype associated with more than 180 features including (most typically) characteristic facial features, congenital heart defects, and abnormalities of the palate (McDonald-McGinn et al., 1999). Parenting a child with any developmental disability presents unpredictable challenges (Carroll, 2013; Rolland & Walsh, 2006), likely to increase the carer's risk of mental health problems such as depressive symptoms and anxiety ((Hartling et al., 2014; Miodraga & Hodapp, 2010; Singer, 2006). However, the plight of parents caring for a child in adult life with 22q11DS is poorly understood, in part due to the large inter- and intra-familial symptomatic variability (Shprintzen, 2008). Additionally, and despite the unique variability in 22q11DS, there is no research that highlights the individual articulated account of parenting a child with 22q11DS into adult life. Therefore this study aims to explore the 'lived' experience of parenting a child with 22q1DS into adult life. It explores both positive and negative subjective interpretations of the unique phenomenon from the parents' perspective.

The behavioural phenotype of 22q11DS is characterised by intellectual disability and/or learning problems, and specific cognitive impairments including executive dysfunction (Bish, Ferrante, McDonald-McGinn, Zackai, & Simon, 2005), attention deficits (Niklasson, Rasmussen, Oskarsdóttir, & Gillberg, 2005), and social impairment (Shashi et al., 2012). Comorbidity with autism spectrum disorders (Fine et al., 2005), anxiety disorders (Fung et al., 2010), mood (Green et al., 2009), and psychotic disorders is high compared with the general population (Murphy, Jones, & Owen, 1999). In adult life, a higher rate of unemployment compared to those without disabilities (Sanford et al., 2011) will inhibit

financial independence, creating relational and financial complications for both older parents and the adult child.

Due to the variable nature of 22q11DS, poor public awareness, and ignorance in many health professionals, parents of children and adults with 22Q11DS are poorly supported in their complex lives. Largely ignored in mainstream psychological research, inferences can only be made on their likely experience of disenfranchised grief, or grief/loss that is not recognised or validated by others (Doka, 1989), a plethora of sad emotions on receiving the diagnosis (as reported in Hallberg, Óskarsdóttir, & Klingberg, 2010) and intense disappointment for the child they desired or expected. Their own life dreams invariably and necessarily are placed on hold or adjusted as they assume a lifetime of care for a child who is unlikely to reach total independence. For some, traumatic responses to health related events that threaten the life of their child may be cumulative and complex. When chronic illness and disability is part of a parenting experience, there is likely shock at the diagnosis, long hospital stays forcing separation from the child, fear of disability and/or death, and reduced quality of life.

Trauma symptoms in parents who have a child with chronic or critical illnesses such as cancer and accidental injury are well-recognised (e.g., Brocque, Hendrikz, & Kenardy, 2010; McCarthy, Ashley, Lee, & Anderson, 2012). For instance, 83% of parents whose child underwent cardiac surgery before the age of 3 months exhibited evidence of experiencing at least one trauma response at a clinical level (Franich-Ray et al., 2013). Conversely, if medical treatment and hospitalization is experienced as traumatic by the child, difficulties can arise relationally with parents viewed as unintended accomplices of the traumatic event (Stuber & Shemesh, 2006). Therefore, it is possible that parents who have a child with 22q11DS also experience traumatic responses from both primary and vicarious exposure.

Despite psychological risks, there is a growing body of research that highlights the possibility of positive psychological changes as a result of a struggle with adversity (Joseph & Linley, 2008). With the rise of positive psychology in the early 1990s, the construct of growth out of adversity, or posttraumatic growth, has come to define distress as a catalyst for positive psychological change (Joseph & Linley, 2005; Tedeschi & Calhoun, 1996). Growth out of adversity is defined as a positive change in psychological functioning after trauma, such as developing strengths, changing life values and beliefs, and accepting personal limitations (Joseph, 2012).

Joseph and Linley's (2005) organismic valuing process theory purports that a traumatic event can shatter an individual's former worldview causing traumatic distress. Psychological growth out of such adversity will only occur however, if the individual is able to integrate the new trauma-related information into a new world view (Joseph & Linley, 2005). Additionally, if the social environment can provide the human needs of autonomy, competence, and relatedness, then growth will be promoted (Ryan & Deci, 2000). However, complicating recovery, integration of the trauma related information can take a positive or negative pathway occurring in one of three ways: a) assimilation of the experience, returning to pre-trauma baseline but remaining vulnerable to future traumas (e.g. I'm invincible), b) negative accommodation of the experience causing psychopathology (e.g. bad things happen to me), or c) positive accommodation of the experience leading to growth (e.g. I can learn from this).

Psychological growth as a result of parenting a child with a developmental disability has been recorded in the literature. For example, families have been found to adapt and thrive in the face of adversity, constructing meaningful stories surrounding the journey with their child (e.g., Green, 2002; King et al., 2006; Rolland & Walsh, 2006). Although parents may grieve the child they expected, they can discover new pathways of happiness or spirituality as

a result of having a child with a developmental disability (e.g., King et al., 2006; Myers, Mackintosh, & Goin-Kochel, 2009; Retzaff, 2007). No research highlights whether parents experience these life-changing epiphanies as a function of having a child with 22q11DS.

This interpretative, phenomenological study explores subjective interpretations of parents who have raised a child with 22q11DS into adult life. It seeks both positive and negative 'lived' interpreted experiences of experiencing this unique disability from a parental perspective. Interpretative Phenomenological Analysis (IPA; Smith, 1996), underpinned by phenomenology, double hermeneutics, and symbolic interactionism, is a suitable qualitative methodology for this uniquely 'lived' experience (Smith, 2004) as it seeks idiographic meaning making. As such, this study explored parental sense making of 22q11DS from the perspective of: a) parenting; b) experience of support; c) perception of 'self' change over time, and d) expectations of their future influenced by their child's disability and related experiences.

Method

Participants

Eight parents (two male, six female) of an adult child with 22q11DS were recruited from a supporting foundation. Participants formed a homogenous, purposive sample relating to the unique phenomenon under investigation (Smith & Osborn, 2008). Only parents who themselves did not have 22q11DS were included in the study. The demographic characteristics of participants and their children are outlined in Table 1. Pseudonyms are used to protect the participants' identities. All eight participants were interviewed individually with two sets of couples contained in the sample. George and Gabriella are married to each other, as are Max and Maria.

Insert Table 1 here

Procedure

Following university human ethics clearance, recruitment occurred through an online support group. Additionally, letters were sent to parents who had participated in previous research at the University conducting this study and had consented to be notified regarding future studies. Potential participants were screened for eligibility following contact with the researcher. Prior to the interview, study materials (i.e., participant information statement, consent form, and outline of the semi-structured interview) were sent to the participants.

Data was collected through open-ended, semi-structured interview questions that allowed for reiterative sense-making of the participants' rich, personal accounts (Smith, 2011; Smith & Osborn, 2008). Interviews were conducted one-on-one at the participant's home, with the exception of two interviews completed via telephone at the request of the participants. All interviews were digitally audio-recorded and ranged in duration from 52 to 155 minutes. Participants were reimbursed for their time with a $20 gift card.

Epistemology

The philosophical underpinnings and methodological approach of the current study were based on phenomenology, symbolic interactionism, and hermeneutics. That is, we aimed to capture the constructed meaning around the phenomenon of having an adult child with 22q11DS by engaging the participants in reflective interpretation of their experience (Blaikie, 1991). Meaning-making is formed through interactions. As such, symbolic interactionism refers to the participants' sense-making which is created as a result of social interaction, modified through individual interpretations (Smith, 1996). Further, as environments (and thus interpretations) vary, participants attribute their own subjective meanings to the experience of having an adult child with 22q11DS. However, as the researchers' access to the participants' personal world is affected by their own conceptions, a double hermeneutic is involved. That is, the researcher strives to make sense of the participant making sense (Smith & Osborn, 2008).

Analysis

Rigor in qualitative research demands ongoing verification undertaken through step by step guidelines to ensure reliability and validity. Transparency occurred through conducting procedures as described by Smith et al. (2009). Thus, interviews were audio-recorded and transcribed verbatim by the first author. Independent auditing was conducted by the first and second authors each developing an audit trail from reading and re-reading transcripts, noting relevant items and psychological constructs in the margin. The subjective interpretation of each author gradually proceeded from descriptive to interpretative (Smith, Jarman, & Osborn, 1999). The first and second authors then engaged in robust discussion to identify and agree on relevant convergent and divergent themes supported by rich data both within and across the data set. A summary of higher order themes and subthemes were grouped into clusters under appropriate headings. Finally, a narrative analytic account was used to link theory to themes generated through pertinent verbatim extracts from transcript. Saturation is not sought in IPA given the focus on divergent (one data set) and convergent (all data sets) rich themes (Smith, 1996). Conclusions were drawn in terms of the similarities and differences among participants (see Table 2).

Insert Table 2 here

Credibility

This IPA study sought validity through being credible, and reliability through being dependable. This meant that through a process of reflexivity, the researchers continued to question personal and professional experiences (stated below) and how these affected the interpretation of the study. Personal experiences, logs of independent analysis, and open discussion of biases brought debate and an acknowledgement of our own subjective, constructed world as limiting the research. For example, the absence of explicit interpretation of trauma by participants provided considerable challenge for the authors not to

force the data. Dependability occurred by subjecting the analysis to IPA as described above (Smith & Osborn, 2008). Rather than a post-hoc assessment of worthiness, the audit trail accounted for the systematic examination at each level of analysis (e.g., transcripts, independent audits, meetings, notes, tracking between authors). The auditing authors wrote the analysis bringing further transparency to final themes.

Author's perspective

All of the authors are current researchers in disability and trauma within family life. The second author has worked for over two decades in this context as a therapist. Each researcher was conscious of the need to bracket biases to guard against forcing the data into preconceived interpretations surrounding disability. However, the authors were similarly conscious of the importance of knowledge and experience in this field of enquiry for engaging with the unexpected through independent audits, the audit trail, and reflection through discussion and write-up at all stages.

Insert Table 3 here

Results

One superordinate theme: *Losing 'I'; Finding 'self';* overarches six subordinate themes: (1) *Stigma and a double-edged sword; (2) Where is 'I'; (3) Conflicting loss, grief, and guilt; (4) Angry advocacy; (5) Pragmatic acceptance;* and (6) *Finding authenticity and purpose*. These themes highlight the participants' descriptions, such as the isolation and stigma seeping into their lives as they progressively lose their own independence, friendships, and instinctual judgement. Battling against hierarchical structures that are suspicious of their actions and intent, angry advocacy pushes back against layers of obstruction and ignorance. Diagnosis is a double-edged sword of relief and grief, demanding they co-journey an unchartered pathway with often indifferent professionals. Finding 'self' comes through a pragmatic re-evaluation of 'accomplishments' that allows an authentic and positive

psychological shift in their whole-of-life interpretation. As such, psychological growth is experienced through conscious engagement with empathy, humility, gratitude and pride.

Stigma and a double-edged sword

This theme explores the stigmatisation experienced as hovering throughout their child's life. Self-questioning and blame associated with *why their child has this disability* remains a lurking burden on these participants. As a consequence, guilt is a well-rehearsed response to perceived judgement by health professionals, particularly prior to the diagnosis. The 'not knowing' leaves them with the perception that they are the target of judgement; unfairly laden with confusion and guilt:

> They were very critical as to what I did or what I ate during my pregnancy or what medication I took … they make you feel really uncomfortable. [Tracy]

Here we can see Tracy describes the syndrome in terms of the widespread stigma, inclusive of school, where these participants struggled at numerous levels to educate against ignorance in staff and other parents. Participants sensed little understanding or empathy of their journey. It continues to be exhausting and irksome. Again, without open communication, suspicions of judgement left them without a voice, adding to the feeling of infectious stigma:

> I don't know whether they thought the syndrome could be caught like a disease… You're at school but you're on the perimeter … we were just singled out and singled out on so many occasions. [Tracy]

For some, obvious and immediate signs of disability are perceived to bring open support. For those whose children do not immediately appear to be disabled, fear of invalidation is never far from the surface. There is a sense that these participants oscillate between the relief of their child being not quite disabled enough, and the frustration of *'not quite there'* for support and assistance:

> That's the difficulty – that she's not quite there, but she's not down there either. [Gabriella]

Participants watch hopelessly as their children slip between the cracks of much needed support. Feeling embattled, these participants continue to promote their own perceptions of their child's needs. Stigma and suspected disbelief from professionals has inherently burdened them throughout the journey:

> For really disabled people I think there's possibly stuff but for someone like him who just looks fully functioning …it's hard. Every two years he has to have a letter that says he's got a disability … It's genetic (laughs)…it's not going to go away. [Sandra]

Prior to the diagnosis, participants were unsure whether to trust their instincts, and oscillated between judgement and concern about their child's development. Mostly, they sensed relief when the label of 22q11DS was given. The diagnosis provided validation for the parents' concerns. It relieved some guilt, and initially gave the parents new-found confidence in their intuitive skills:

> It was quite an event because we knew what she had… There was an answer as to why. [Gabriella]

However, diagnosis proved a double-edged sword, as initial relief was replaced with sorrow that the child has a syndrome that brings life adversity. Lost was the hope that would not give way to doubt prior to diagnosis. As demonstrated in Sandra's quote below, receiving a label of 22q11DS can cause more uncertainty than not having a diagnosis at all:

> It's nice to have a diagnosis. But then you look at the list of signs and symptoms and you think what sort of diagnosis is that?! [Sandra]

For many, a diagnosis of 22q11DS was unchartered territory where concern and uncertainty, variation in symptomatology, and a medical model that promotes professionals as experts had to be confronted. Powerlessness, head butting, and incompetence became the

new modus operandi. Tracy highlights the struggle of having parenting knowledge validated among medical professionals who were unfamiliar with the syndrome:

> You really felt like you were hitting your head on a brick wall. We've been there, done that, it doesn't work. Doctors don't like you to tell them that… [Tracy]

Where is 'I'?

These parents lost themselves in their child as they became the managers of their child's health, behaviour, and extensive needs. Most are running a family and working. Feeling all-consumed and exhausted, they constantly crave respite that never comes, as formal support is lacking:

> You've just got to keep on looking after your family. You don't get time out, you don't get any super answers, or anything like that. [Wendy]

There are few variations on this theme. Whether it is the exhaustion, or the sense that this is unending; all participants perceived themselves as caught in a lifelong career of care for children who cannot attain independence. Dramatic ultimatums are sometimes the only way to coerce assistance in their exhausted state of worry:

> We were so tired … we said (to the doctor), "Either you give her the tablets, or we're going to take them!" [Maria]

Participants openly expressed fear for their child's fate once they can no longer provide all the necessary care. For them, it is not a matter of indispensability, but of commitment and love. Guilt was never far from their musings on who will care when they are no longer alive. They were unable to imagine others offering the care their child requires:

> What's going to happen? It's a bloody great concern, great concern…It's a big ask to say look, if something goes wrong, it's your job. [Max]

Throughout the journey, the participants' stated that their relationships changed. The parents' self is entangled with their child, and their child's pain is transferred to them. There

is still disappointment at the perceived disinterest directed at their child from once valued friends:

> Adults wouldn't take time to talk to her…and that I found…very disappointing. No compassion even in my close friends. [Maria]

Lack of knowledge about the syndrome from professionals was interpreted as a personal insult. There was the sense that Anna's value of self struggles against the judgement she perceives from others:

> It kind of makes me feel like…we're worthless…that's how it feels, when they don't know about it - makes me feel like I'm just rubbish. [Anna]

Conflicting Loss, Grief, and Guilt

This theme captures the life participants expected prior to having a child with 22q11DS. They see the potential trajectories of their life, and feel sorrow and grief for what could have been. Although they appeared generally happy with the path they have been given, there were cautious reflections on the 'other' life, which they perceived as being a simpler and more naïve existence, without the extreme challenges they have experienced. Participants struggled against self-pity with each milestone that should have passed when reflecting on the losses and unendingness that 22q11DS brings. Sandra reflected on this loss:

> There's all those things that change…no wedding, no grandchildren…live with him for the rest of your life… [Sandra]

The parents did not only grieve for themselves, they dreamt of how they could change their child's fate:

> I think, if you just had these few bits of chromosome, a few bits of DNA, you'd probably be something really extraordinary. And it's sad. [Max]

Female participants continued to mourn their desired mothering experience, remembering the cloak and dagger around having a sick child. Pain still appeared raw as they reflected on the child's birth, and their inability to celebrate their new motherhood:

> That's supposed to be a really joyous time…We couldn't get the baby baptised, and our friends couldn't see her … we had this phantom baby. [Maria]

Birth memories of loss remain poignant, even though their child is now an adult. This is a different commitment to the one they imagined. Rather than a journey of positive surprises, they cared for a uniquely sick baby, and had the burden of unknowing. Isolated and alone, social connections dwindled:

> That's where the social isolation started … You join a 1% club, with 1% of people with a sick baby. [Maria]

Like any adult, the children with 22q11DS have hopes for their future. Participants speak of bringing their child back to reality when dreams exceed the child's capabilities. While trying to spare their child from the pain of failure, participants are forced to play the 'bad guy' and remind the child of their limits. Conflict is common, and rather than stretching the umbilical cord of independence, these participants are trapped into reining in their child and crushing dreams:

> She doesn't understand that she doesn't actually have the ability… so we've had a lot of tears. [Wendy]

Participants recognise that they are engaging their child in a charade of independence, adjusting tasks to fit their child's competence. They seem trapped in the need to give their child the illusion that they are living a normal adult life. Conflict is evident in the participants as they recognise their collusion with 'normality' on the one hand, and self- criticism on the other, because they are not being true to their goals of teaching their children to work around their challenges:

> She has got her own business … but it's hardly flourishing…She's running it in our house so whenever she's got a client we've got to be as quiet as a mouse. [George]

Similarly, the confusion of protection is complicated by the surreptitious guilt that nags them: *did we do something wrong?* Despite knowing their child's deletions are de novo, the participants are not able to disengage from self-responsibility:

> It's probably just something that weighs on your conscience … I've often wondered about it … if we could've avoided all this had we known better? [George]

Challenges for the child due to 22q11DS magnified parental pain. Guilt turns inward and self-blame is like a volcano simmering under the surface. Anna verbalised the rumination present in all the participants:

> Did we do something wrong … I just feel like, why couldn't it be me, why couldn't I have had all the problems and not my son … it just makes you feel like a failure. [Anna]

Participants compensate for their guilt by enabling their child to achieve which seems to be a strategy for reducing their own distress, by guarding against situations that could upset the child. As their child grows and seeks more independence, they speak of the struggle to relinquish control and let the child make their own mistakes. Should they take the easy path, knowing they will be called on to pick up the pieces when things do not go to plan; or should they stick to the higher goals of learning from mistakes?:

> We've probably always tried to take the protective role but now that she's older it's not working… but I think really afterwards she's realised that – well you were right. [Gabriella]

Angry advocacy

Throughout the journey of having a child with 22q11DS, participants are constantly thwarted in their attempts to manage their child's condition, particularly by the experts treating them. Frustration is never far from the surface, ever ready to defend against the professional that excludes them or dismisses their insight:

> This woman came into the house and said, "Get your cardiologist to tell you what he's not telling you…she's got VCFS." [Maria]

The lack of empathy from professionals coupled with uncertainty surrounding the condition leaves participants feeling powerless. Max feels blocked at each turn and senses the indifference to his questions, creating more questions and concerns:

> There's a huge range of symptoms…so where are we? Dunno … What's the prognosis? Dunno. What causes it? Dunno … What do we do now? Dunno. What happens in the long term? Well, a few develop schizophrenia … So how can we tell? You can't. You don't know until you get there. [Max]

Anna finds the obstruction particularly difficult, as it compounds the guilt she feels. She lost time that could have been used to help her son. Anna shifts between exasperation with medical professionals' secrecy regarding their lack of understanding about 22q11DS, and anger with them for not informing her more:

> That's what hurts. They knew nothing about the syndrome and they were treating him … Why couldn't they tell us that's going to happen? [Anna]

Pragmatic acceptance

A diagnosis of 22q11DS often brings more questions than answers. Initially, participants search for answers. They then learn that questioning is futile, and become resigned to the uncertainty that casts a shadow on their life. Participants avoid feeling victimised by asking themselves, 'why *not* me?':

> If there's 1/100 births that has a heart condition, then why shouldn't it be us? Someone's got to make up those numbers, so…it was us! [Wendy]

Now their children are adults, participants feel a lot of the difficulties (e.g., struggling through school) are behind them, and have made an uneasy peace with the ambiguity of their child's future. They recognise that they are not in control of the overall journey and seem to consider anxiety as fruitless and a waste of time:

> There'll be a few more bumps…I'm not too concerned, although I really have no idea what the future brings, so… definitely say a lot of Hail Marys…it's got me this far, I'm sure it'll get me through a few more years. [Tracy]

Re-evaluation of their life values and expectations emerges as a direct reflection of having a child with a disability. Dreams they had for their expected child are now abandoned, and there is a momentary collision with disappointment and sadness about missed opportunities:

> That's what changes the most - is your expectation of life … and that's disappointing. [Sandra]

However, their child's version of success is considered more openly, and delight is embraced for what they have achieved. Accomplishments of any type trigger a fierce pride in these participants as they accept their child's life path may deviate from the traditional. Value and successes worthy of celebration are adjusted:

> I would not stop talking about our kids … I wouldn't cover it up; I wouldn't hide it. [Sandra]

Finding authenticity and purpose

Participants' knowledge about the syndrome and themselves appears to have been a dual journey of growth. The long search for answers for themselves has been replaced by a wish to pass on their own knowledge and experience. When opportunities for altruism arise, participants volunteer. They hope they can make the journey easier for others. The participants know how difficult it can be to have an adult child with a developmental disorder, and express frustration when others are perceived as not advocating for equality when they have the potential. In particular, George is angered by this:

> I've recognised the difficulties that people with disabilities have in making their way in life … When I could do stuff I did…because I was the only person who seemed to have any commitment to it. [George]

The participants are redefining their identity through a recognition that everybody in the family has benefitted from the child with 22q11DS. There is a sense of belonging. Again, participants see the path life they could have taken without a child with 22q11DS. Instead of mourning what could have been, they are grateful they have become the parent they are:

> I've had the big transformation. I'm really pleased with that because…looking back…I don't like the mother I would have been. [Maria]

Other participants make sense of the diagnosis because they 'know' they have innate gifts that enable them to cope with the immense strain of having a child with a developmental disorder. They philosophically engage with being given what we can bear and belief they were 'given' their child because they are strong and positive. Perseverance when times are tough is perceived as an existential gift:

> God's given the hard troubles to me because he knew I wouldn't pike out. [Anna]

Discussion

This study highlighted the unremitting burden experienced by these participants in parenting a child with 22q11DS. Overarching these results is the uninvited intrusion of their child's extensive needs. Their narratives define the journey as one of *Losing 'I'* and ultimately *Finding 'self'*. The subthemes expose the cumulative stressors experienced by these participants that continually change and often become more threatening as their child ages. Physical and mental exhaustion is felt as a constant in their lives. Stigma, in particular, brings isolation to these once independent adults. Progressively they lose their adult friendships, and lack trust in their instinctual judgement as they are doubted and questioned over their child's presentation to professionals. Though they find advocacy skills to battle hierarchical structures, anger is a part of that armoury that pushes back against layers of obstruction and ignorance.

For these participants, eventual diagnosis was a mix of relief and grief as they entered an unchartered pathway with often indifferent professionals and abdicated from the imagined

parenting experience they had expected. Each participant spoke of the creative challenge to redefine their lives as they confronted an uncertain future for their adult child with little societal assistance. Finding 'self' comes through a pragmatic re-evaluation of theirs and their child's 'accomplishments' that allows an authentic and positive psychological shift in their whole-of-life interpretation. As such, the flexibility to redefine self brings a conscious engagement with psychological wellbeing, mirrored in the growthful domains of empathy, humility, gratitude and pride.

In contrast to previous research highlighting the potential for trauma in parents with ill children (e.g. Franich-Ray et al., 2013), these participants did not directly express being traumatised by their parenting of a child with 22q11DS. However, hypervigilance was noted as cumulative on earlier disappointment and it stretched their creative risk assessments to offset potentially traumatising events. Instead of trauma, participants speak of the burden and frustration of snowballing adversity throughout their child's life, which is described as feeling stigmatised, shamed and self-doubting. Grief and loss are palpable in these participants as their child misses milestones and is actively excluded. These are similar to the feelings expressed in a qualitative study of mothers who had multiple births, with at least one of the children affected by disability (Bolch, Davis, Umstad, & Fisher, 2012). For example, contrasting their own experiences with mothers who had healthy children brought about significant distress (Bolch et al., 2012).

The current study demonstrates the parents' disenfranchised grief (Doka, 1989) which has also been reported in parents of children with special needs (Bolch et al., 2012) and people who have received genetic testing results for themselves and their families (Sobel & Cowan, 2003). Participants experienced sorrow and loss throughout their child's life and were unprepared for the stigmatisation of disability and the aloneness such stigma brought. The grief surrounding their child and related experiences cannot be openly acknowledged or

publicly mourned because they are not recognised or validated due to this ever-present stigma (Doka, 1989) and the poor profile of this disability in society. Medical professionals and educators contributed to this; either through ignorance or a dismissive attitude towards parents' concerns. This is a clear avenue for intervention, as both these groups are well-placed to normalise the reactions to having a child with 22q11DS. Parents who have a child with 22q11DS experience complex and conflicting emotions. Reducing institutional stigma can provide these parents with an avenue to seek support and thus lessen the social isolation described.

These interviews were in many ways alive with shifts in perception and interpretation occurring throughout. There was a sense that their future was evolving before them with uncertainty. As they spoke of that future in which they would no longer be able to care for their child, there was a profound sadness and sense of exhaustion, as they planned extensively without societal support for the future care of their adult child. The anticipatory loss and trauma casts a shadow over their everyday lives as it does for many individuals who live their daily life in anticipation of adversity (McCormack, White, & Cuenca, 2016). However, as they reflected on the journey, they began to interpret the stigma, ignorance, loss of relationships, and battles through layers of obstruction as providing the springboard for psychological growth through reconnecting with their empathetic and grateful self, and honouring pride in their journey. Redefining meaning in their experiences brought reconciliation with past belief systems to combat helplessness, frustration, and stigma; comparable to people who received predictive genetic testing for Huntington Disease (Sobel & Cowan, 2003).

The psychological growth experienced by these participants is similar to previous research surrounding positive aspects of having a child with a developmental disability; where parents have reported outcomes including finding new perspectives on life, increased

sensitivity, opportunities to learn, improved family dynamics, and increased confidence and assertiveness (Hastings & Taunt, 2002). For example, parents with a child affected by autism or Down syndrome have been found to positively adapt by examining and adapting their views surrounding their child and their parenting role, even though grief is still real for the dreams that are no longer attainable (King et al., 2006). Viewing these positive perceptions (or 'psychological growth out of adversity' as we have conceptualised it) as a coping resource is valuable in terms of helping parents to adapt (Hastings & Taunt, 2002). As evidenced by the current study, conflicting interpretations can be experienced simultaneously (e.g., loss and gain).

Time can be an important factor in adjusting positively to parenting a child with a disability (Krauss & Seltzer, 1993). For example, although not defined as psychological growth, early studies also found that older mothers of developmentally disabled adults had similar or better outcomes on measures of depression, life satisfaction, parenting stress, and social isolation compared to a) older women not in a caregiving role, b) women who were caregivers for older adults and c) young mothers who had a child with an intellectual disability (Krauss & Seltzer, 1993). Taylor and Seltzer (2011) also found that the mother-child relationship improved while children with autism were in high school. The fact that the participants in the current study all had adult children may have contributed to the more positive outcomes these parents experienced, such as recognising their innate gifts as carers. As parents, they have experienced grief, loss, and guilt throughout their child's life; all of which may have caused self-reflection and thus psychological growth. Future research should aim to delineate which factors are best predictive of these positive outcomes, and how best they can be promoted and supported in families affected by disability. For example, a heterogeneous group in terms of parents' age and years since diagnosis could be compared to parents with adult children.

Limitations

As a qualitative study this research does not seek to generalise, nor seek cause and effect. Double hermeneutics played an important role in the analytic process of this unique phenomenon, therefore the researchers' experiences and biases may have impacted both positively and negatively on their interpretations. In an attempt to enhance the study's credibility, the auditors vigorously adhered to an ongoing audit trail inclusive of multiple robust discussions. Although the sample may not be representative of all parents who have an adult child with 22q11DS as there are many factors impacting these relationships, the use of IPA has sought to provide hypotheses for future and larger research through an in-depth exploration of a homogenous group's subjective interpretations that have experienced parenting a child with 22q11DS. Therefore, the findings contribute to the body of knowledge surrounding 22q11DS by highlighting both positive and negative interpreted impacts on these parents, particularly as their children moved into adulthood.

Conclusions

This study draws attention to the many struggles faced by parents of an adult child with 22q11DS and the potential for psychological growth, especially their ability to redefine 'self' and embrace an authentic way of living with 22q11DS. Importantly, the coexistence of distress and psychological growth is a major consideration for therapists, health care workers, and support personnel to work in more creative ways with families caring for adult children with disabilities. Growthful domains of empathy, humility, gratitude and pride are likely triggers for psychological wellbeing despite ongoing distress and loss. Of interest is that the challenges from health care services and feelings of guilt appeared to trigger growth in these participants. This provides a basis for further qualitative and quantitative research and for aims of therapeutic support.

It is important that medical professionals acknowledge any gaps in their own knowledge when presented with unusual medical phenomenon. In doing so, opportunities for open dialogue can occur assisting rapport for supporting parents with realistic expectations. Supporting parents on a life-long journey of care is a relational challenge for many medical personnel. Though positive and negative outcomes are part of the journey with 22q11DS, the attitude of the health professional can impact parents' reactions to their child's disability. Family relations, parental age, years from diagnosis, and expectations on siblings are considerations for future research, support, care policies and programmes and factors for consideration in larger quantitative research.

Clinicians can access recently published guidelines for managing adults with 22q11DS (Fung et al., 2015), guiding them to provide appropriate care for the person affected by 22q11DS, and for managing and validating parents' concerns for their child. Following the guidelines is imperative during the affected person's adolescence and adulthood, where complications such as schizophrenia can arise (Murphy et al., 1999). Medical professionals also have the opportunity to offset stigma, blame, and self-doubt; the common legacies of disability for many families. There is guidance available for clinicians that can aid in this process, with advice on communicating effectively with parents and families from diagnosis and throughout their journey (see Kisler & McConachie, 2010). This can encourage new and diverse interpretations of their complex experiences, and subsequently support psychological growth out of the adversity of 22q11DS.

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Transcript Extract Notations

— A pause in speech.

… Removal of nonessential material.

Table 1

Participant and child characteristics

Participant				Child				
	Gender	**Age**	**Marital status**	**Gender**	**Age**	**Age of diagnosis**	**Developmental ability***	**IQ**
'Anna'	F	54	Married	M	24	10 months	2	60 or below
'Gabriella'	F	63	Married	F	28	8 years	5	71-80
'George'	M	63	Married					
'Maria'	F	58	Married	F	23	3 years	5	74
'Max'	M	62	Married					
'Sandra'	F	55	Married	M	29	7 years	4	110
'Tracy'	F	58	Married	M	25	3.5 years	5	60-65
'Wendy'	F	57	Married	F	21	2 years	5	71-80

Note. * is the child's developmental ability as rated by the parents on a scale of 1 -7, where 1 = severely delayed, 7 = not delayed at all.

Table 2

Stages of Interpretative Phenomenological Analysis

Stage	Description
1	Re-listening, transcription, reading and re-reading.
2	Developing emergent themes through independent interpretation (e.g., noting).
3	Credibility established through robust author discussion.
4	Repeating stages 1 – 3 for the other seven cases.
5	Searching for connections across emergent themes, identifying convergence and divergence.
6	Clustering of subordinate themes 'Stigma and a double-edged sword', 'Where is 'I', 'Conflicting loss, grief, and guilt', 'Angry advocacy', 'Pragmatic acceptance', and 'Finding authenticity and purpose' that support the superordinate theme 'Losing I; Finding Self'.
7	Reviewing transcripts to validate interpretations in the results.

Table 3

Summary of themes

Superordinate theme: Losing 'I'; Finding 'self'	
Subordinate themes	**Description**
Stigma and a double-edged sword	Stigma invades the participants' lives because the 'not knowing' leaves them the target of judgement, or victims of indifferent professionals. The diagnosis is a double-edged sword of relief and grief.
Where is 'I'	Participants lose themselves in the management of their child's health, behaviour, and needs. There is no respite from this role, and as such relationships change.
Conflicting loss, grief, and guilt	The participants cautiously reflect on the life that could have been without 22q11DS and mourn each milestone that should have passed. Although logically they know their child's deletions are de novo, they wonder if they did something to cause it.
Angry advocacy	Participants battle against hierarchical structures suspicious of their actions and intent. They must fight the layers of obstruction and ignorance to receive the care their child needs.
Pragmatic acceptance	There is an uneasy peace with the ambiguity of their child's future. Participants re-evaluate their expectations and learn to celebrate success for their child.
Finding authenticity and purpose	Psychological growth is experienced through conscious engagement with empathy, humility, gratitude and pride.